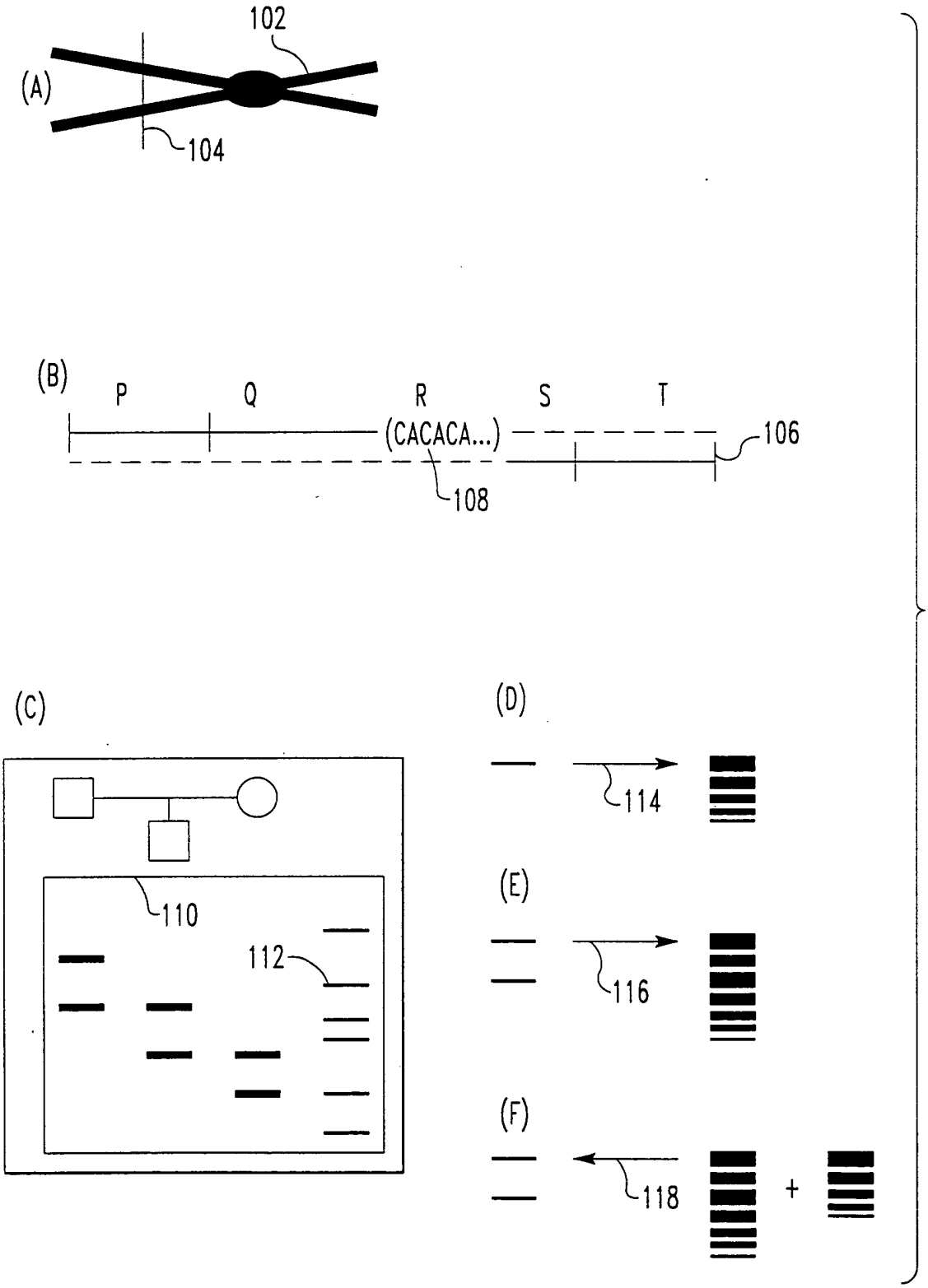

FIG. 1A

(STEP 1) ACQUIRE AN INDIVIDUAL'S GENOMIC DNA

(STEP 2) PERFORM PCR AMPLIFICATION AT AN STR LOCUS OF THIS DNA

(STEP 3) SIZE SEPARATION ASSAY OF THE AMPLIFIED PCR PRODUCTS

(STEP 4) ANALYZE THE PEAKS OF THE RESULTING ASSAY INTO DNA SIZE VS. CONCENTRATION FEATURES

(STEP 5) DECONVOLVE THE ANALYZED PCR PRODUCT TO DETERMINE THE GENOTYPE OF THE INDIVIDUAL AT THE STR LOCUS

(STEP 5') DECONVOLUTION USING FOURIER DOMAIN SIGNAL PROCESSING

(STEP 6) EMPLOYING A PCR STUTTER PATTERN LIBRARY

FIG. 1B

US 6,338,931 B2 Jan. 15, 2002

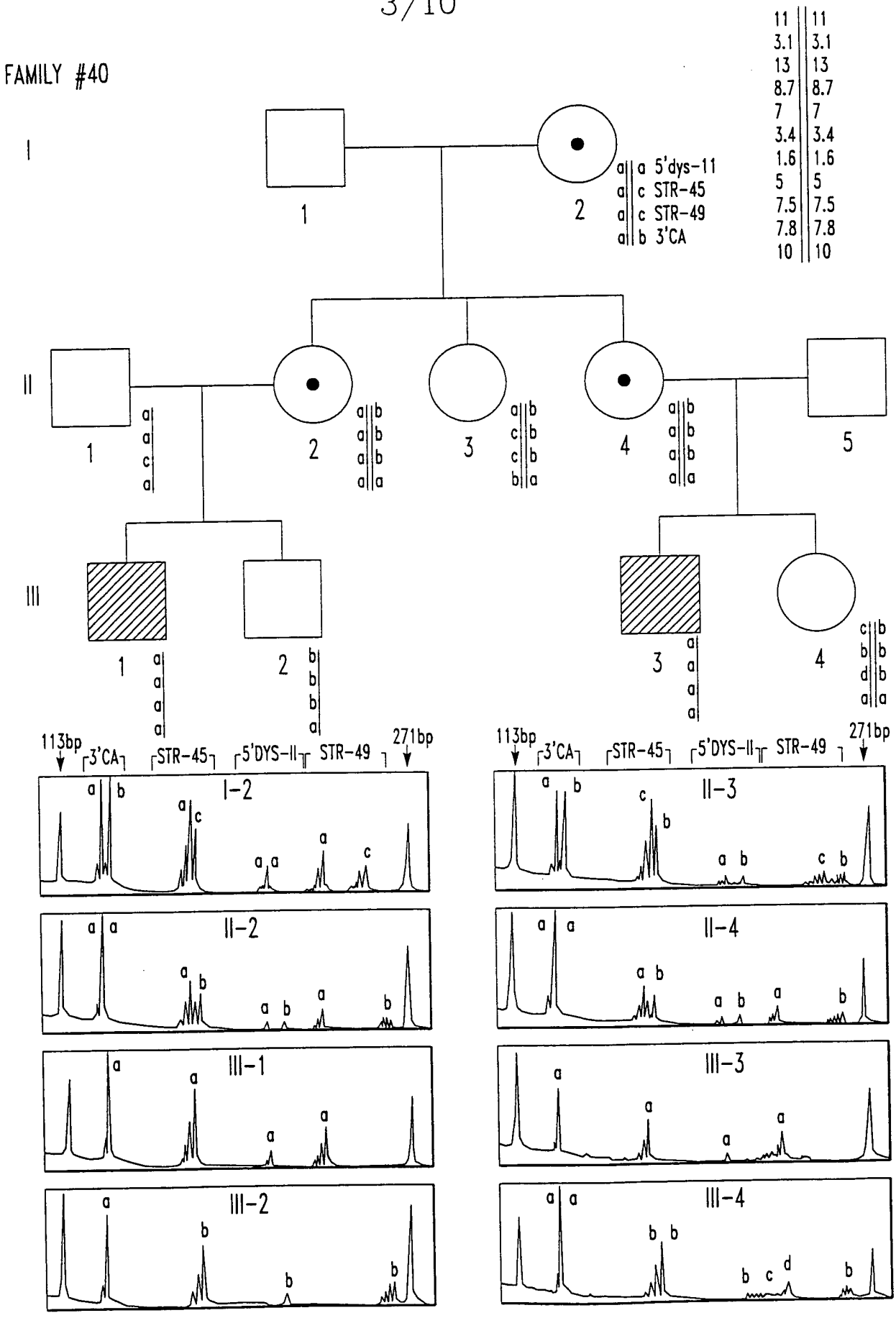
FAMILY #40
I
1
2
a a 5'dys-11
a c STR-45
a c STR-49
a b 3'CA
11 11
3.1 3.1
13 13
8.7 8.7
7 7
3.4 3.4
1.6 1.6
5 5
7.5 7.5
7.8 7.8
10 10
II
1 a a c a
2 a a a a / b b b a
3 a c c b / b b b a
4 a a a a / b b b a
5
III
1 a a a a
2 b b b a
3 a a a a
4 c b d a / b b b a
113bp
3'CA
STR-45
5'DYS-II
STR-49
271bp
I-2
II-2
III-1
III-2
II-3
II-4
III-3
III-4

FIG.2

DATA FROM MARKER STR-45.

SIZE	INDIVIDUAL A	INDIVIDUAL E
161	821	930
163	2171	1928
165	7242	5896
167	20799	18115
169	55373	47391
171	101299	94852
173	0	61326
175	0	0

DATA FROM MARKER STR-49.

SIZE	INDIVIDUAL D
221	843
223	1217
225	2360
227	6123
229	11469
231	26811
233	48135
234	0
236	0
238	0
240	0
242	0
244	0
246	0
248	0
250	0
252	1695
254	2877
256	5410
258	11553
260	17482
262	25866
264	28672

FIG. 3A

USING THE MW MARKERS TO CONSTRUCT THE DATA EXPECTATIONS

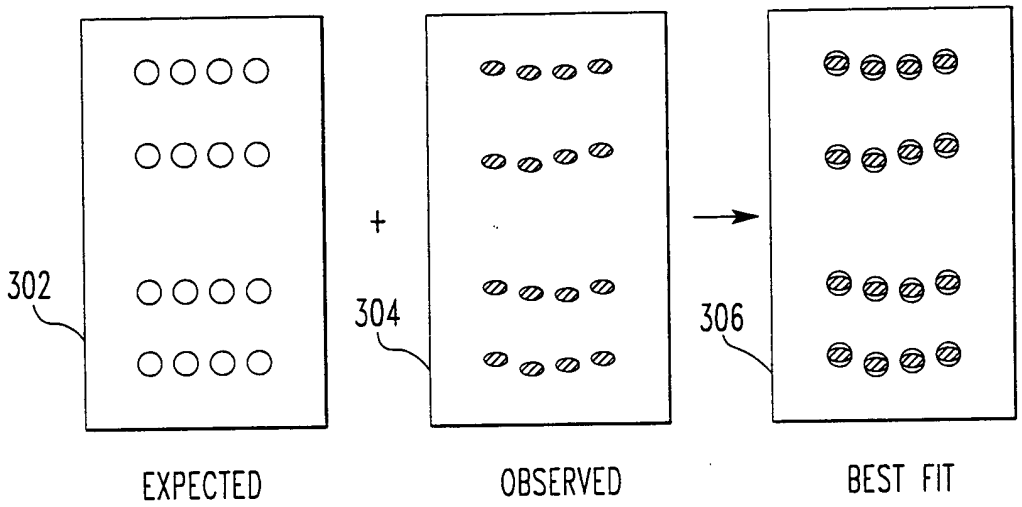

FIG.3B

USING THE DATA EXPECTATIONS TO LOCALIZE AND QUANTITATE DATA

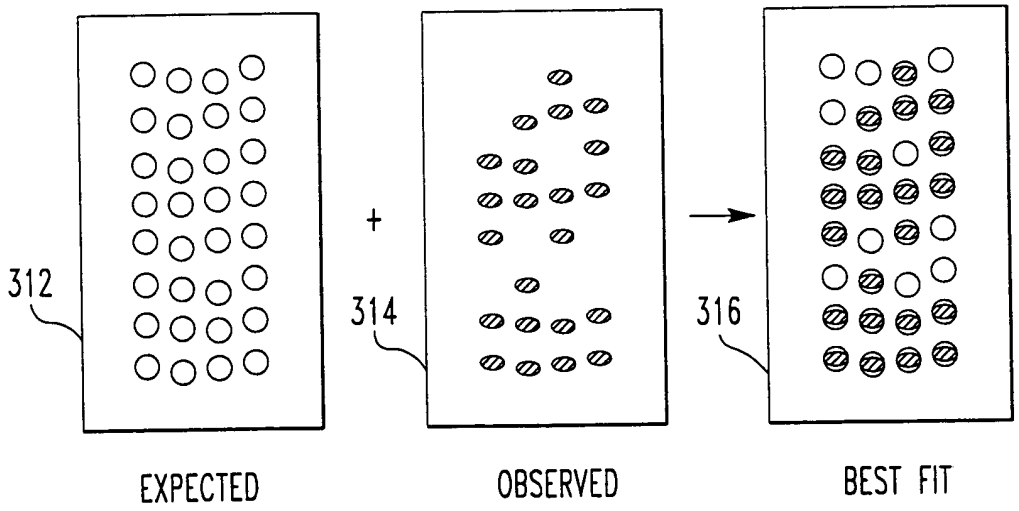

FIG.3C

09/02/2005 - 03:03:02

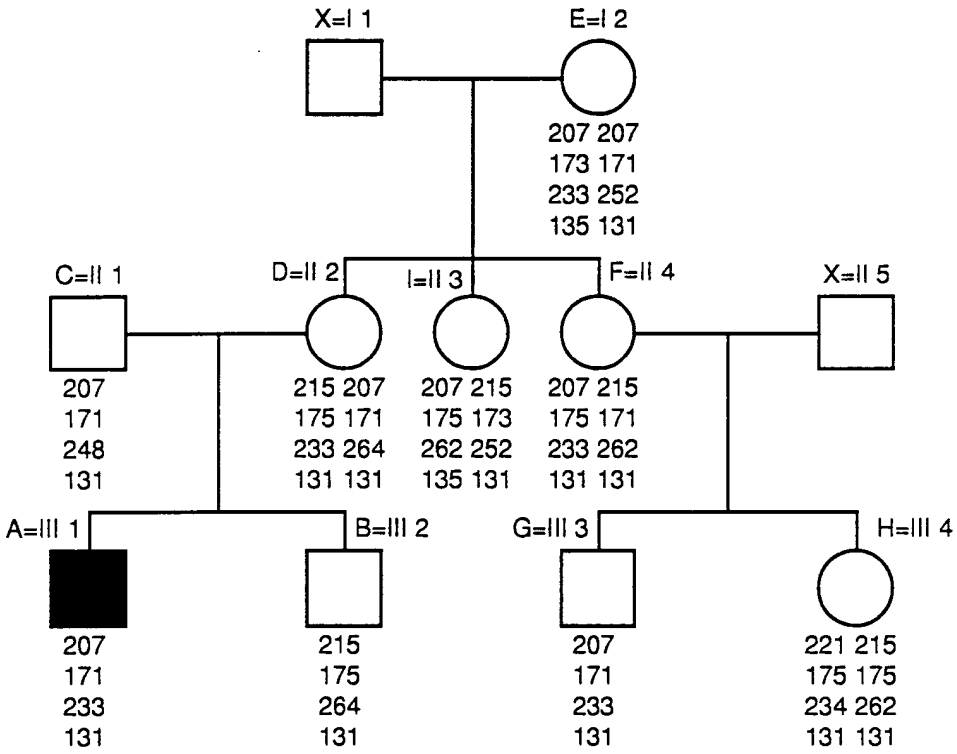
X=I 1
E=I 2
207 207
173 171
233 252
135 131
C=II 1
D=II 2
I=II 3
F=II 4
X=II 5
207
171
248
131
215 207
175 171
233 264
131 131
207 215
175 173
262 252
135 131
207 215
175 171
233 262
131 131
A=III 1
B=III 2
G=III 3
H=III 4
207
171
233
131
215
175
264
131
207
171
233
131
221 215
175 175
234 262
131 131

FIG. 4

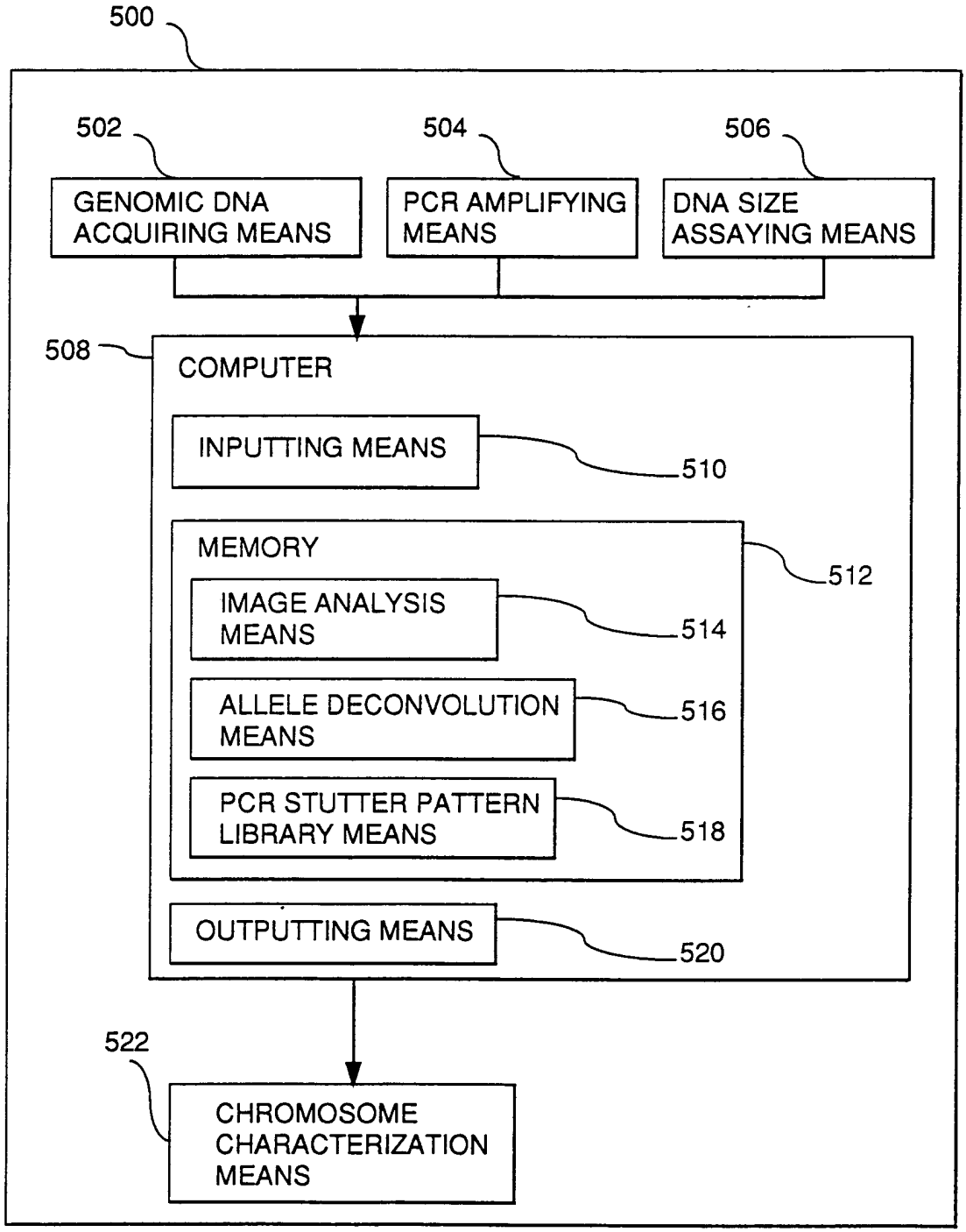
500
502
GENOMIC DNA ACQUIRING MEANS
504
PCR AMPLIFYING MEANS
506
DNA SIZE ASSAYING MEANS
508
COMPUTER
INPUTTING MEANS
510
MEMORY
512
IMAGE ANALYSIS MEANS
514
ALLELE DECONVOLUTION MEANS
516
PCR STUTTER PATTERN LIBRARY MEANS
518
OUTPUTTING MEANS
520
522
CHROMOSOME CHARACTERIZATION MEANS

FIG. 5

(STEP 1) DETERMINE GENOTYPES OF RELATED INDIVIDUALS.

(STEP 2) SET CHROMOSOME PHASE BY GRAPH PROPAGATION, DEDUCTIVE METHODS, OR LIKELIHOOD ANALYSIS.

(STEP 3) DETERMINE THE PHENOTYPIC RISK OF DISEASE FOR THE INDIVIDUALS.

(STEP 4) PRESENT THE RESULTS.

FIG. 6

SUBSTITUTE SHEET (RULE 26)

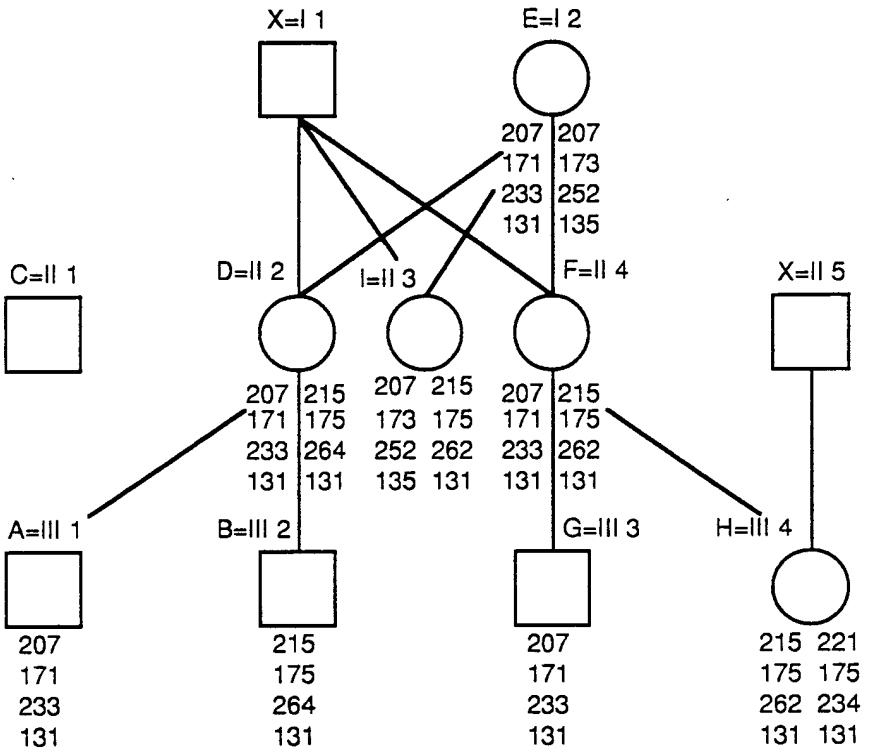
X=I 1
E=I 2
207 207
171 173
233 252
131 135
C=II 1
D=II 2
I=II 3
F=II 4
X=II 5
207 215
171 175
233 264
131 131
207 215
173 175
252 262
135 131
207 215
171 175
233 262
131 131
A=III 1
B=III 2
G=III 3
H=III 4
207
171
233
131
215
175
264
131
207
171
233
131
215 221
175 175
262 234
131 131

FIG. 7

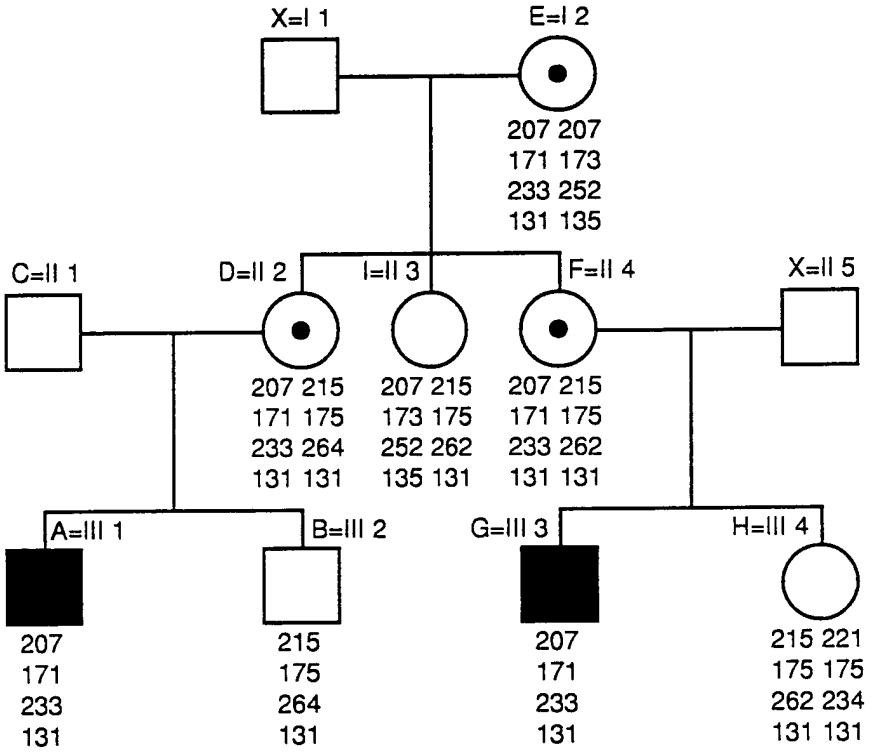
X=I 1
E=I 2
207 207
171 173
233 252
131 135
C=II 1
D=II 2
I=II 3
F=II 4
X=II 5
207 215
171 175
233 264
131 131
207 215
173 175
252 262
135 131
207 215
171 175
233 262
131 131
A=III 1
B=III 2
G=III 3
H=III 4
207
171
233
131
215
175
264
131
207
171
233
131
215 221
175 175
262 234
131 131

FIG. 8